General

Guideline Title

European Respiratory Society guidelines for the diagnosis of primary ciliary dyskinesia.

Bibliographic Source(s)

Lucas JS, Barbato A, Collins SA, Goutaki M, Behan L, Caudri D, Dell S, Eber E, Escudier E, Hirst RA, Hogg C, Jorissen M, Latzin P, Legendre M, Leigh MW, Midulla F, Nielsen KG, Omran H, Papon JF, Pohunek P, Redfern B, Rigau D, Rindlisbacher B, Santamaria F, Shoemark A, Snijders D, Tonia T, Titieni A, Walker WT, Werner C, Bush A, Kuehni CE. European Respiratory Society guidelines for the diagnosis of primary ciliary dyskinesia. Eur Respir J. 2017 Jan 4;49(1):1-25. PubMed

Guideline Status

This is the current release of the guideline.

This guideline meets NGC's 2013 (revised) inclusion criteria.

Recommendations

Major Recommendations

The strength of the recommendations (strong, weak) and levels of evidence (high, moderate, low or very low) are defined at the end of the "Major Recommendations" field.

Evidence-Based Recommendations for the Use of Each of the Six Tests Considered for Primary Ciliary Dyskinesia (PCD) Diagnosis

Clinical Features

Which patients should be referred for diagnostic testing?

Based on moderate confidence in the evidence:

1. The Task Force recommends that patients are tested for PCD if they have several of the following features: persistent wet cough; situs anomalies; congenital cardiac defects; persistent rhinitis; chronic middle ear disease with or without hearing loss; a history in term infants of neonatal upper and lower respiratory symptoms or neonatal intensive care admittance (strong recommendation).
2. Patients with normal situs presenting with other symptoms suggestive of PCD (as listed in recommendation 1 above) should be referred for diagnostic testing (strong recommendation).
3. Siblings of patients should be tested for PCD, particularly if they have symptoms suggestive of PCD (as listed in recommendation 1) (strong recommendation).
4. The Task Force recommends the use of combinations of distinct PCD symptoms and predictive tools (e.g., PICADAR) to identify patients for diagnostic testing (weak recommendation).

Nasal Nitric Oxide

In patients suspected of having PCD should nasal nitric oxide be used as a diagnostic tool?

Based on moderate confidence in the evidence, the Task Force recommends that:

1. Nasal nitric oxide measurement should be used as part of the diagnostic work-up of school children aged >6 years and adults suspected of having PCD, preferably using a chemiluminescence analyser with a velum closure technique (strong recommendation).
2. In children aged <6 years suspected of having PCD, the Task Force suggests nasal nitric oxide measurement using tidal breathing as part of the diagnostic work-up (weak recommendation).
   Remark: The Task Force suggests that patients presenting with a strong clinical history should undergo further testing, even if nasal nitric oxide is normal (weak recommendation).

High-speed Video Analysis (HSVA)

In patients suspected of having PCD should HSVA be used as a diagnostic tool?

Based on low confidence in the evidence, the Task Force recommends:

1. HSVA, including ciliary beat frequency and beat pattern analysis, should be used as part of the diagnostic work-up of patients suspected of having PCD (weak recommendation).
2. Ciliary beat frequency should not be used without assessment of ciliary beat pattern in diagnosing PCD (strong recommendation).
3. To improve diagnostic accuracy of HSVA, ciliary beat frequency/pattern (CBF/P) assessment should be repeated after air–liquid interface (ALI) culture (strong recommendation).

Transmission Electron Microscopy (TEM)

In patients suspected of having PCD should TEM be used as a diagnostic tool?

Based on low confidence in the evidence, the Task Force recommends:

1. Ciliary ultrastructure analysis by TEM should be used as part of the diagnostic work-up of patients suspected of having PCD (strong recommendation).
2. Further diagnostic investigations should be performed in patients with normal ultrastructure if the clinical history is strong (strong recommendation).[#]
3. In patients with hallmark ciliary ultrastructure defects for PCD further confirmatory diagnostic investigations are not required (strong recommendation).[¶]

[#]Normal ciliary ultrastructure, as resolvable by TEM, does not exclude the diagnosis of PCD (16% of PCD-positive patients have TEM without a detectable defect).

[¶]Patients with hallmark ciliary ultrastructure defects for PCD (absence of outer dynein arms, combined absence of inner and outer dynein arms, inner dynein arm absence combined with microtubular disarrangement) assessed by TEM almost always have PCD (false-positive results are very rare ≈0.7%).

Genetics

In patients suspected of having PCD, should genotyping be used as a diagnostic tool?

There were no studies that fulfilled inclusion criteria to answer this question. Statements to assist the clinician are made in the genetics sections of the original guideline document but these are not evidence based. Therefore, the Task Force could not make formal recommendations as for other diagnostic procedures. However, the Task Force has provided a list of Task Force statements on genetics, which is based upon agreement between experts rather than published evidence.

Immunofluorescence

In patients suspected of having PCD, should immunofluorescence be used as a diagnostic tool?

There were no studies that fulfilled inclusion criteria to answer this question. Statements to assist the clinician are made in the immunofluorescence sections of the original guideline document but these are not evidence based. Therefore, the Task Force could not make formal recommendations as for other diagnostic procedures. However, they have provided a list of Task Force statements on immunofluorescence, based upon agreement between experts rather than published evidence.

Definitions

Quality of Evidence

Using the GRADE (Grading of Recommendations Assessment, Development and Evaluation) approach, the Task Force rated the overall quality of evidence for each question as high, moderate, low or very low, based on the following criteria: (1) study design, (2) risk of bias, (3) directness, (4) consistency, (5) precision and (6) publication bias. Criteria 2-6 are assessed as either serious or very serious. Grading of the evidence as HIGH, MODERATE, LOW or VERY LOW was based initially on the study design and then downgraded appropriately based on the other factors.

Strength of Recommendations

The final grading of the evidence helped to inform the final recommendations as either STRONG (should always be done) or WEAK (should be performed in certain circumstances). For reaching recommendations, the Task Force took into account the quality of the evidence; the balance between benefits and harms; the patients' values and preferences and other factors such as costs, feasibility, accessibility etc.

Clinical Algorithm(s)

A diagnostic algorithm for primary ciliary dyskinesia (PCD) is provided in the original guideline document.

Scope

Disease/Condition(s)

Primary ciliary dyskinesia (PCD)

Guideline Category

Diagnosis

Clinical Specialty

Family Practice

Internal Medicine

Medical Genetics

Otolaryngology

Pulmonary Medicine

Intended Users

Advanced Practice Nurses

Physician Assistants

Physicians

Respiratory Care Practitioners

Guideline Objective(s)

To provide evidence-based recommendations on diagnostic testing for primary ciliary dyskinesia (PCD), especially in light of new developments in such tests, and the need for robust diagnoses of patients who might enter randomised controlled trials of treatment

Target Population

Patients referred for primary ciliary dyskinesia (PCD) testing in whom the PCD diagnosis was either confirmed or excluded

Interventions and Practices Considered

1. Assessment of clinical features (signs and symptoms) of primary ciliary dyskinesia (PCD)
2. Nasal nitric oxide (NO) measurement
3. High-speed video microscopy
4. Genetic analysis (genotyping)
5. Assessment of ciliary structure with transmission electron microscopy
6. Immunofluorescence analysis of protein mislocalisation

Major Outcomes Considered

Sensitivity and specificity/accuracy of diagnostic tests

Methodology

Methods Used to Collect/Select the Evidence

Searches of Electronic Databases

Description of Methods Used to Collect/Select the Evidence

Systematic Review

Literature Search Methods

The Task Force searched the OVID Medline and EMBASE databases using the search terms outlined in supplementary table 2 (see the "Availability of Companion Document" filed]) to address each PICO (Patient, Intervention/Investigation, Comparison, Outcome) focussed question. In a first step, at least two researchers from each Work Group (WG) screened the titles and abstracts, to exclude manuscripts that clearly did not address the PICO or the WG's additional questions. In a second step, two searchers (one for genetics due to lack of researchers) reviewed the full texts of the remaining papers, to identify manuscripts that addressed the PICO and fulfilled the inclusion criteria. Third, the committee and WG members received the lists of identified papers and were asked to report any additional studies not identified by the search. All data fulfilling the *a priori* inclusion criteria were included.

The Task Force included all peer reviewed manuscripts from 1996 to 14th March 2016 with no language limitations. It was decided that manuscripts predating 1996 would be unlikely to reliably diagnose primary ciliary dyskinesia (PCD) versus non-PCD according to current standards. The Task Force excluded conference proceedings, grey literature and studies in non-humans.

Number of Source Documents

- Clinical features: 8 studies were included in qualitative synthesis; 2 studies directly addressed PICO (Population, Intervention, Comparison, Outcome question).
- Nasal nitric oxide: 23 studies were included in qualitative synthesis; 4 studies directly addressed PICO.
- High speed video: 30 studies were included in qualitative synthesis; 2 studies directly addressed PICO.
- Transmission electron microscopy (TEM): 19 studies were included in qualitative synthesis; 11 studies directly addressed PICO.

- Genetics: 95 studies were included in qualitative synthesis; no studies directly addressed PICO.
- Immunofluorescence (IF): 41 studies were included in qualitative synthesis; no studies directly addressed PICO.

See flow charts (Figure 1 a-f) in the supplementary material (see the "Availability of Companion Documents" field) for the process of identification and inclusion of studies.

Methods Used to Assess the Quality and Strength of the Evidence

Weighting According to a Rating Scheme (Scheme Given)

Rating Scheme for the Strength of the Evidence

Quality of Evidence

Using the GRADE (Grading of Recommendations Assessment, Development and Evaluation) approach, the Task Force rated the overall quality of evidence for each question as high, moderate, low or very low, based on the following criteria: (1) study design, (2) risk of bias, (3) directness, (4) consistency, (5) precision and (6) publication bias. Criteria 2-6 are assessed as either serious or very serious. Grading of the evidence as HIGH, MODERATE, LOW or VERY LOW was based initially on the study design and then downgraded appropriately based on the other factors.

Methods Used to Analyze the Evidence

Review of Published Meta-Analyses

Systematic Review with Evidence Tables

Description of the Methods Used to Analyze the Evidence

Systematic Review

Data extraction tables were designed to capture information required for each Work Group (WG). These were circulated for editing to the Task Force (TF) panel. Each WG decided what data was required a) to answer the PICO (Patient, Intervention/investigation, Comparison, Outcome), and b) to answer additional questions. Data were extracted by two independent researchers with the exception of genetics WG which used single extraction due to lack of researchers. Since there is no reference standard for diagnosis of primary ciliary dyskinesia (PCD), details of how diagnosis was confirmed/excluded was extracted for all studies and acceptability agreed by the TF panel.

Quality of Evidence Leading to Recommendations

Grading of Recommendations Assessment, Development and Evaluation (GRADE) is a method for systematically assessing the quality of evidence for a diagnostic test and then making recommendations for use of the test based on the quality of this evidence. Using the GRADE approach the TF rated the overall quality of evidence for each question as high, moderate, low or very low, based on the following criteria: study design, risk of bias, directness, consistency, precision and publication bias.

The identified manuscripts were assessed on the following criteria –

1. Study design – for example a randomised controlled trial (although very few exist in diagnostics) would be a higher level of evidence than prospective cohort studies and these would be higher than case-control studies.
2. Risk of bias – The TF assessed risk of bias using the Quadas-2 tool for the quality assessment of diagnostic accuracy studies, based on four domains (a) patient selection; b) conduct or interpretation of index test; c) selection, conduct or interpretation of reference standard; and d) patient flow.
3. Directness - This refers to the existence of a direct link between the diagnostic test and patient important outcomes. For intervention studies, intermediate outcomes, such as accuracy of diagnostic tests, are always considered "indirect" evidence and thus reduce the quality. Therefore, directness was graded as "potentially serious" in all WGs.
4. Consistency - This refers to the degree to which reported study results (e.g., sensitivity, specificity) from included studies are similar; thus heterogeneity of results was reported as inconsistency.

5. Precision – Precision refers to the degree of certainty concerning the estimates of each test performance (quantified by the width of confidence intervals around estimates).
6. Publication bias – This indicates that studies may have been published selectively and pooled estimates of published studies might not reflect the truth (e.g., negative findings have not been published, or are unavailable).

Criteria 2-6 are assessed as either serious or very serious. Grading of the evidence as High, Moderate, Low or Very Low was based initially on the study design and then downgraded appropriately based on the other factors.

Methods Used to Formulate the Recommendations

Expert Consensus (Delphi)

Description of Methods Used to Formulate the Recommendations

Task Force and Work Group Composition

The membership and roles of the Task Force (TF) panel are summarised in Supplementary Table 1 (see the "Availability of Companion Documents" field). A leadership group of four were responsible for chairing meetings, providing support to the work groups and monitoring progress. This leadership group also coordinated the writing of the practice guideline and oversaw the editing. Work Group (WG) leaders were proposed and agreed at the first meeting of the task force, based on their expertise. Following training from European Respiratory Society (ERS) methodologists in Grading of Recommendations Assessment, Development and Evaluation (GRADE), systematic reviewers drafted protocols for the searches, conducted systematic reviews, extracted data from the chosen manuscripts, assessed the quality of the data and finally synthesised the data using narrative and if appropriate meta-analysis.

The TF panel comprised experts and trainees in the field of primary ciliary dyskinesia (PCD) from multidisciplinary backgrounds including pulmonologists; ear, nose, and throat (ENT); cell scientists; electron microscopists and geneticists. Their expertise included clinical phenotyping, screening tests including nasal nitric oxide (nNO), ex-vivo and in-vivo ciliary function tests including high-speed video microscopy analysis (HSVA) and radioaerosol mucociliary clearance, transmission electron microscopy (TEM), cell culture (submerged and at air-liquid interface [ALI]), lung physiology and imaging, epidemiology and qualitative research. Some members of the panel lead national diagnostic centres, and there were members from countries where diagnostic facilities are limited. Members of the panel volunteered to participate in WG activities based on their expertise and interests. The ERS provided support to the panel from two methodologists, an advisor for dissemination and a junior committee member; the methodologists did not participate in the votes of the recommendations, the dissemination advisor and junior committee member were paediatric pulmonologists and did contribute to WG activities, panel discussions and voting.

A larger group with interest in PCD has met annually at ERS Congresses since 2006. The opinions of this group of over 60 clinicians, nurses, scientists and allied health professionals were sought and taken into account when deciding which tests to evaluate and which questions needed answering by the TF.

Two patient representatives participated in the first task force meeting, helped in the project design, contributed to the writing of the practice guideline and the dissemination of the report. The European Lung Foundation contributed to the first meeting. An international survey and semi-structured interviews were conducted to understand the patient perspective.

Formulation of the Topics and Questions

The panel met with a wider group of professionals (n=80) interested in PCD during ERS Congress 2014. A semi-structured discussion led to understanding of current diagnostic pathways and tests across Europe, and the questions that clinicians and scientists need answering. These discussions informed a closed meeting of the TF panel. The panel agreed that six diagnostic tests (clinical symptoms, nasal nitric oxide [nNO], high speed video-microscopy [HSV], transmission electron microscopy [TEM], genotype and immunofluorescence labelling of ciliary proteins [IF]) would be evaluated using a 'PICO' (Patient, Intervention/Investigation, Comparison, Outcome) structured question: "Patients suspected of having PCD, Investigated by nNO, TEM etc., when Comparing patients with a final positive or negative diagnostic outcome, what was the diagnostic accuracy (Outcome) of the test?" The TF primarily aimed to identify studies of consecutive patients referred for PCD testing, in whom the PCD diagnosis was either confirmed or excluded. In the absence of sufficient literature of this study design, it was agreed that the comparator group might include healthy controls, or patients with other respiratory diseases (e.g., cystic fibrosis [CF], asthma) from case control studies, but this would down grade the level of evidence. Lack of a gold standard diagnostic test for PCD was a limitation for this project. Diagnostic performance indicators (e.g., sensitivity and specificity) were therefore compared to the authors' final decision regarding positive/negative diagnosis based on available tests. The PICO questions for each test were finalised during several rounds of teleconferences and email discussions (see Table S2 in

supplementary materials).

Several less structured questions were agreed to provide the basis of a narrative synthesis, but these questions were not used to provide recommendations.

Quality of Evidence Leading to Recommendations

The final grading of the evidence helped to inform the final recommendations as either STRONG (should always be done) or WEAK (should be performed in certain circumstances). For reaching recommendations, the Committee took into account the quality of the evidence; the balance between benefits and harms; the patients' values and preferences and other factors such as costs, feasibility, accessibility etc. Evidence profiles were discussed with and across WGs electronically and by telephone conferences throughout the duration of the TF and discussed in a face-to-face meeting of the entire TF panel at the 2015 ERS Congress in Amsterdam. Sections of the manuscript were written by WG leaders and members of their groups, and again discussed and amended electronically across WGs and within the committee. Evidence that was of a lower quality than that used for recommendations was commented on in the guideline but was not used to make recommendations.

Consensus Statement for Diagnostic Outcomes

The TF conducted a modified Delphi survey in four rounds to develop consensus regarding the contributions of diagnostic tests to confirm or refute a diagnosis of PCD. Only members of the Task Force with relevant expertise participated by completing online questionnaires (https://www.isurvey.soton.ac.uk/). Respondents were anonymous to others with the exception of the Chair who could identify participants. Before each round participants reviewed the results of previous surveys, including a summation of comments with reasons underlying opinions and recommendations for iterations. The first round of the survey aimed to understand if any individual tests could definitively confirm or exclude a diagnosis of PCD. In the second round each Delphi participant was asked to review the summary of responses from round 1; they were then invited to consider combinations of tests that might confirm or exclude a diagnosis when the diagnosis is considered clinically very likely, or only modest. In round 3 and 4 there were further iterations. A consensus was reached when 80% of participants were in agreement.

Rating Scheme for the Strength of the Recommendations

Strength of Recommendations

The final grading of the evidence helped to inform the final recommendations as either STRONG (should always be done) or WEAK (should be performed in certain circumstances). For reaching recommendations, the Task Force took into account the quality of the evidence; the balance between benefits and harms; the patients' values and preferences and other factors such as costs, feasibility, accessibility, etc.

Cost Analysis

A formal cost analysis was not performed and published cost analyses were not reviewed.

Method of Guideline Validation

Internal Peer Review

Description of Method of Guideline Validation

These guidelines were endorsed by the European Respiratory Society (ERS) Science Council and Executive Committee in September 2016.

Evidence Supporting the Recommendations

Type of Evidence Supporting the Recommendations

The type of supporting evidence is identified and graded for each recommendation (see the "Major Recommendations" field).

Benefits/Harms of Implementing the Guideline Recommendations

Potential Benefits

The patient representatives to the Task Force fully endorsed that an accurate diagnosis was an important outcome, because it leads to a better recognition of their problems by physicians and more effective treatment, and thus improves their health and quality of life. This was confirmed by a questionnaire survey of 352 PCD patients from 25 countries and 20 in-depth interviews.

Potential Harms

False-positive or false-negative diagnosis

Qualifying Statements

Qualifying Statements

- The guidelines published by the European Respiratory Society (ERS) incorporate data obtained from a comprehensive and systematic literature review of the most recent studies available at the time. Health professionals are encouraged to take the guidelines into account in their clinical practice. However, the recommendations issued by this guideline may not be appropriate for use in all situations. It is the individual responsibility of health professionals to consult other sources of relevant information, to make appropriate and accurate decisions in consideration of each patient's health condition and in consultation with that patient and the patient's caregiver where appropriate and/or necessary, and to verify rules and regulations applicable to drugs and devices at the time of prescription.
- The patient representatives to the Task Force fully endorsed that an accurate diagnosis was an important outcome, because it leads to a better recognition of their problems by physicians and more effective treatment, and thus improves their health and quality of life. However, diagnostic accuracy studies do not provide direct evidence for the improvement of patient-important outcomes; consequently, the confidence in results of test accuracy studies can be judged, at best, as moderate.

Implementation of the Guideline

Description of Implementation Strategy

An implementation strategy was not provided.

Implementation Tools

Clinical Algorithm

For information about availability, see the *Availability of Companion Documents* and *Patient Resources* fields below.

Institute of Medicine (IOM) National Healthcare Quality Report Categories

IOM Care Need

Living with Illness

IOM Domain

Effectiveness

Patient-centeredness

Identifying Information and Availability

Bibliographic Source(s)

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Adaptation

Not applicable: The guideline was not adapted from another source.

Date Released

2017 Jan 4

Guideline Developer(s)

European Respiratory Society - Professional Association

Source(s) of Funding

European Respiratory Society

Guideline Committee

European Respiratory Society Guidelines for the Diagnosis of Primary Ciliary Dyskinesia Task Force

European Respiratory Society Guidelines for the Diagnosis of Primary Ciliary Dyskinesia Work Groups

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Financial Disclosures/Conflicts of Interest

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Disclosure of Conflicts of Interest

Panel members disclosed potential conflicts of interest according to European Respiratory Society (ERS) policies at the start of the Task Force and prior to publication of this manuscript. Following review of these statements, the Chairs and ERS Guidelines committee considered it unnecessary for any panel member to abstain from decisions for any of the recommendations.

Conflict of Interest

D. Rigau and T. Tonia are employees of the ERS. Other disclosures can be found alongside this article at erj.ersjournals.com

Guideline Status

This is the current release of the guideline.

This guideline meets NGC's 2013 (revised) inclusion criteria.

Guideline Availability

Available from the European Respiratory Journal Web site .

Availability of Companion Documents

The following is available:

- European Respiratory Society guidelines for the diagnosis of primary ciliary dyskinesia. Online supplement. 58 p. Available from the European Respiratory Journal Web site .

Patient Resources

None available

NGC Status

This NGC summary was completed by ECRI Institute on April 7, 2017. The information was verified by the guideline developer on May 2, 2017.

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